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Brief Clinical Report

New MCA/MR Syndrome With Generalized Hypotonia, Congenital Hydronephrosis, and Characteristic Face

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We have observed a newly recognized syndrome in two unrelated Japanese patients. Manifestations include severe mental retardation, growth failure, generalized floppiness, congenital hydronephrosis, cardiac anomalies, cleft palate, and characteristic face. To date, caused genesis is unknown. Am. J. Med. Genet. 68:347–349, 1997.

KEY WORDS: MCA/MR syndrome; congenital hydronephrosis; cleft plate

INTRODUCTION

We have observed a newly recognized MCA/MR syndrome in two unrelated patients. We describe the differential diagnosis.

CLINICAL REPORT

Patient 1

The patient was a 2-year-old girl born to nonconsanguineous healthy Japanese parents. Her brother was normal. Fetal ultrasonography showed congenital hydronephrosis. At term, birth weight was 2,800 g (20th centile), length was 47 cm (20th centile), and head circumference (OFC) was 32.5 cm (35th centile). Examination showed coarse face with midface hypoplasia, synophrys, long eyelashes, prominent eyes, low nasal bridge, short nose, submucosal cleft palate, open mouth, excessive nuchal skin fold, and webbed neck (Fig. 1a). Her ear lobes were large and asymmetric. She had mild hypertrichosis on her back and limbs. Her joints were loose and hypermobile (Fig. 1b). Overlapping toes were noted. Echocardiography showed malformed aortic valves with mild stenosis and regurgitation; VSD, ASD, and PDA had closed. Urological investigations showed bilateral hydronephrosis due to stenosis of ureteropelvic junction. Vesicoureteral reflux was noted. She had repeated urinary tract infections. Karyotype was normal. No major CNS anomaly was demonstrated by CT and MRI of the brain. Weight was 7.7 kg (<1st centile), length was 77 cm (<1st centile), and OFC was 43 cm (<1st centile) at the age of 30 months. She was floppy and could not support her head. Her mental development was at the level of early infancy. Deep tendon reflexes were decreased. Ocular following was poor.

Patient 2

The patient was a 15-month-old boy who was born at term to non-consanguineous healthy Japanese parents not related to patient 1. His brother was normal. Fetal ultrasonography showed congenital hydronephrosis. At 35 weeks of gestation, his birth weight was 2,084 g (35th centile). Examination showed manifestations similar to those of patient 1. He had cleft palate, prominent eyes, short nose, open mouth, short webbed neck, and excessive nuchal skin fold (Fig. 2a). He had mild hypertrichosis of the face and back. Generalized floppiness was noted (Fig. 2b). Echocardiography documented endocardial cushion defect. Urological investigations showed bilateral hydronephrosis due to stenosis of ureteropelvic junction. No major CNS anomaly was demonstrated by CT scan of the brain. Karyotype was normal. Weight was 5.4 kg (<1st centile), length was 65.6 cm (<1st centile), and OFC was 43.0 cm (<1st centile) at age 15 months. Although he started to roll over at 1 year, head control was not complete. He required tube feeding. Deep tendon reflexes were decreased.

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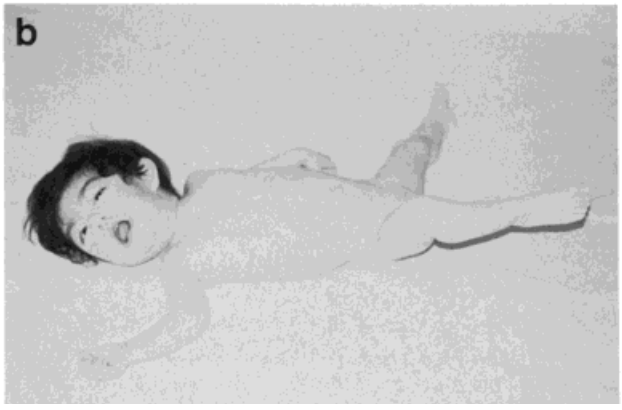

Fig. 1. **a:** Facial appearance of patient 1 at 24 months showing midface hypoplasia, prominent eyes, epicanthus, hypertrichosis of the forehead, synophrys, short nose, ear anomaly, open mouth, and hypotonic facial muscles. **b:** General appearance of patient 1 at 15 months showing generalized hypotonia.

DISCUSSION

Our two unrelated patients are summarized in Table I. Differential diagnosis include Schinzel-Giedion syndrome [Schinzel and Giedion, 1978], the syndrome described by Sakati et al. [1983], neonatal Marfan syn-

Fig. 2. **a:** Patient 2 at 10 months. Facial appearance was similar with that of patient 1. Webbed neck was evident. **b:** Patient 2 at 15 months showing generalized hypotonia.

TABLE I. Summary

	Patient	
Manifestations	1	2
Generalized hypotonia	+	+
Severe mental retardation	+	+
Severe growth failure	+	+
Microcephaly	+	+
Midface hypoplasia	+	+
Hypertrichosis	+	+
Synophyrs	+	–
Long eye lashes	+	+
Prominent eyes	+	+
Epicanthus	+	+
Apparently low set ears	+	+
Long ear lobe	+	+
Flat nasal bridge	+	+
Short upturned nose	+	+
Long philtrum	+	+
Cleft palate	+	+
Open mouth	+	+
Webbed neck	+	+
Hydronephrosis	+	+
Stenosis of ureteropelvic junction	+	+
Cardiac anomalies	+	+
Loose hypermobile joints	+	+
Overlapping toes	+	–
Hyperconvex nails	+	–

drome [Chemke et al., 1984; Schollin et al., 1988], and fetal benzodiazepine syndrome [Laegreid et al., 1989]. However, manifestations in our patients were at variance with these diagnosis. To date, causal genesis is unknown.

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